



## GARS1 gene

glycyl-tRNA synthetase

### Normal Function

The *GARS1* gene provides instructions for making an enzyme called glycyl-tRNA synthetase. This enzyme is found in all cell types and plays an important role in the production (synthesis) of proteins. During protein synthesis, building blocks (amino acids) are connected together in a specific order, creating a chain of amino acids. Glycyl-tRNA synthetase plays a role in adding the amino acid glycine at the proper place in a protein's chain of amino acids.

### Health Conditions Related to Genetic Changes

[Charcot-Marie-Tooth disease](#)

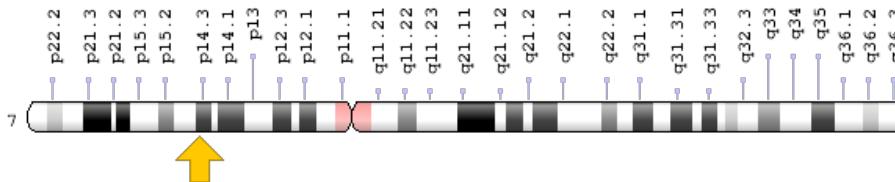
[Distal hereditary motor neuropathy, type V](#)

Several *GARS1* gene mutations have been identified in individuals with distal hereditary motor neuropathy, type V. This disorder affects nerve cells in the spinal cord, resulting in muscle weakness and affecting movement of the hands and feet. The *GARS1* gene mutations that cause distal hereditary motor neuropathy, type V change single amino acids used to make glycyl-tRNA synthetase. It is unclear how *GARS1* gene mutations lead to this disorder. The mutations probably reduce the activity of glycyl-tRNA synthetase. A reduction in glycyl-tRNA synthetase activity may impair transmission of nerve impulses.

## Chromosomal Location

Cytogenetic Location: 7p14.3, which is the short (p) arm of chromosome 7 at position 14.3

Molecular Location: base pairs 30,594,735 to 30,634,033 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CMT2D
- DSMAV
- GARS
- glycine tRNA ligase
- GlyRS
- SMAD1
- SYG\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Specific Enzymes Couple Each Amino Acid to Its Appropriate tRNA Molecule  
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1062>

### Clinical Information from GeneReviews

- GARS-Associated Axonal Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1242>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GARS%5BTIAB%5D%29+OR+%28glycyl-tRNA+synthetase%5BTIAB%5D%29+NOT+%28chromosome+21%5BTIAB%5D%29%29+OR+%28%28CMT2D%5BTIAB%5D%29+OR+%28glycyl+tRNA+synthetase%29+NOT+%2812q24%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## Catalog of Genes and Diseases from OMIM

- GLYCYL-tRNA SYNTHETASE 1  
<http://omim.org/entry/600287>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GARS1.html](http://atlasgeneticsoncology.org/Genes/GC_GARS1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GARS1%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report/#!/hgnc\\_id/HGNC:4162](https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4162)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:2617>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2617>
- UniProt  
<https://www.uniprot.org/uniprot/P41250>

## **Sources for This Summary**

- Antonellis A, Ellsworth RE, Sambuughin N, Puls I, Abel A, Lee-Lin SQ, Jordanova A, Kremensky I, Christodoulou K, Middleton LT, Sivakumar K, Ionasescu V, Funalot B, Vance JM, Goldfarb LG, Fischbeck KH, Green ED. Glycyl tRNA synthetase mutations in Charcot-Marie-Tooth disease type 2D and distal spinal muscular atrophy type V. Am J Hum Genet. 2003 May;72(5):1293-9. Epub 2003 Apr 10.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12690580>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180282/>
- Antonellis A, Lee-Lin SQ, Wasterlain A, Leo P, Quezado M, Goldfarb LG, Myung K, Burgess S, Fischbeck KH, Green ED. Functional analyses of glycyl-tRNA synthetase mutations suggest a key role for tRNA-charging enzymes in peripheral axons. J Neurosci. 2006 Oct 11;26(41):10397-406.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17035524>
- OMIM: GLYCYL-tRNA SYNTHETASE 1  
<http://omim.org/entry/600287>

- Griffin LB, Sakaguchi R, McGuigan D, Gonzalez MA, Searby C, Züchner S, Hou YM, Antonellis A. Impaired function is a common feature of neuropathy-associated glycyl-tRNA synthetase mutations. *Hum Mutat.* 2014 Nov;35(11):1363-71. doi: 10.1002/humu.22681.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25168514>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4213347/>
- James PA, Cader MZ, Muntoni F, Childs AM, Crow YJ, Talbot K. Severe childhood SMA and axonal CMT due to anticodon binding domain mutations in the GARS gene. *Neurology.* 2006 Nov 14;67(9):1710-2. Erratum in: *Neurology.* 2007 Feb 27;68(9):711.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17101916>
- Nangle LA, Zhang W, Xie W, Yang XL, Schimmel P. Charcot-Marie-Tooth disease-associated mutant tRNA synthetases linked to altered dimer interface and neurite distribution defect. *Proc Natl Acad Sci U S A.* 2007 Jul 3;104(27):11239-44. Epub 2007 Jun 26.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17595294>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2040883/>
- Sivakumar K, Kyriakides T, Puls I, Nicholson GA, Funalot B, Antonellis A, Sambuughin N, Christodoulou K, Beggs JL, Zamba-Papanicolaou E, Ionasescu V, Dalakas MC, Green ED, Fischbeck KH, Goldfarb LG. Phenotypic spectrum of disorders associated with glycyl-tRNA synthetase mutations. *Brain.* 2005 Oct;128(Pt 10):2304-14. Epub 2005 Jul 13.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16014653>
- Sleigh JN, Dawes JM, West SJ, Wei N, Spaulding EL, Gómez-Martín A, Zhang Q, Burgess RW, Cader MZ, Talbot K, Yang XL, Bennett DL, Schiavo G. Trk receptor signaling and sensory neuron fate are perturbed in human neuropathy caused by Gars mutations. *Proc Natl Acad Sci U S A.* 2017 Apr 18;114(16):E3324-E3333. doi: 10.1073/pnas.1614557114. Epub 2017 Mar 28.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/28351971>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5402433/>
- Xie W, Nangle LA, Zhang W, Schimmel P, Yang XL. Long-range structural effects of a Charcot-Marie-Tooth disease-causing mutation in human glycyl-tRNA synthetase. *Proc Natl Acad Sci U S A.* 2007 Jun 12;104(24):9976-81. Epub 2007 Jun 1.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17545306>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1891255/>

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